SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA
Azienda Unità Sanitaria Locale di Reggio Emilia

SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA
Azienda Ospedaliera di Reggio Emilia
Arcispedale S. Maria Nuova

Prenatal screening for chromosomal abnormalities through combined test

Information and Informed Consent

Dear Madam,
the Emilia Romagna Region (Emilia Romagna Regional Resolution 533/2008) offers pregnant women the opportunity to take a combined test for the statistical risk assessment of having a child with Down's syndrome (trisomy 21), Edward's syndrome (trisomy 18) and Patau's syndrome (trisomy 13).
Down's syndrome, which is the most common chromosomal abnormality, causes mental disorder and is often associated with cardiac and gastrointestinal abnormalities, as well as hearing and visual disorders. Approximately, one in 700 babies is born with this syndrome.
The likelihood of having a baby with this disease increases as the mother's age increases (general risk).
The combined test allows a risk calculation (specific and individual risk) to be recalculated for every pregnant woman during that pregnancy.

What do we mean by risk?

The risk expresses the chance of an event occurring. For example, a risk of Down's syndrome of 1 in 100 means that there is a 1% chance of having a baby with Down's syndrome.

What is the combined test?

The combined test is one of the methods used to calculate the risk of Down's syndrome and other chromosomal abnormalities. It is a test based on the dose of two substances in the mother's blood and on an ultrasound scan: it allows the likelihood that the foetus is affected by chromosomal abnormalities, especially trisomies 21, 18, 13, to be defined.

How is the combined test performed?

1 – Dual test

It is performed by taking a blood sample from the mother to look at the dose of two substances produced by the placenta (PAPP-A and free-beta-hCG) which in foetuses with chromosomal abnormalities have different values from those found in foetuses with a normal chromosome complement. In our organisation, this sample can be taken between the 9^{th} and 12^{th} week + 5 days of pregnancy. The test is most accurate when the blood sample is taken between the 9^{th} and 10^{th} week of pregnancy, therefore it is advisable to perform it during this period.

2 – Nuchal translucency (NT)

A specific ultrasound scan is performed between the 11^{th} week + 3 days and the 13^{th} week + 3 days of pregnancy to determine the nuchal translucency. The translucency, which is due to a small amount of fluid located in the nape of the foetus between the skin and the underlying tissues, can be measured with an ultrasound scan.

The thickness of the nuchal translucency increases as the pregnancy advances, therefore, a specific value of normal nuchal translucency corresponds to a specific foetal crown-rump length (CRL). When the ultrasound scan detects an increased nuchal translucency thickness, especially over the 99^{th} percentile, the foetus has a higher risk of chromosomal abnormalities (particularly Down's syndrome, and less frequently trisomies 13 and 18), of cardiac defects or rare genetic syndromes, even if the majority of these foetuses will not be affected. In order to perform a correct measurement, the foetal profile must be observed and therefore it may be necessary to wait until the foetus is in the correct position, moving spontaneously, or to prompt movements through light pressure on the mother's abdomen or by asking the patient to cough. This may mean that the test takes longer to complete.

3 - The data is then processed in order to obtain an estimate of the individual risk of trisomies 21, 18 and 13.

Interpretation of the Test

The combined test is a probabilistic test and does not allow full detection or exclusion of a chromosomal abnormality. It must be noted that a negative screening result reduces the risk but it does not completely eliminate it. In other words, in the event of a negative test the presence of chromosomal abnormalities cannot be fully excluded.

In the same way, a positive test does not imply that the foetus is affected by a disease; it only indicates that the risk is high enough to justify an invasive diagnostic procedure. The test is particularly effective for some chromosomal abnormalities, such as trisomies 21, 18 and 13. There are other serious and rarer chromosomal abnormalities which cannot be detected by the combined test. The test is considered positive when the risk is higher than 1/250. In this case, if the patient wants to know for sure if the foetus has a chromosomal abnormality, then she is offered a test to determine the foetal karyotype (i.e. the number and structure of the foetus' chromosomes) through chorionic villus sampling or amniocentesis. These are both invasive procedures and involve an increased risk of miscarriage of about 1%.

Increased nuchal translucency (NT) (i.e. over 95%) can be a risk factor for other foetal diseases. In these cases, a level 2 ultrasound scan combined with a foetal echocardiogram between the 19th and 21st week of pregnancy is appropriate to exclude other abnormalities, particularly malformations.

Finally, we wish to inform you that the ultrasound scan for nuchal translucency is performed by physicians accredited by the Fetal Medicine Foundation (FMF) in London, who are audited in terms of case record and quality of work.

The program used for the risk assessment is also provided by the Fetal Medicine Foundation (FMF) in London, which is the international scientific gold standard for this procedure.

Date .....

Patient's signature

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Physician's stamp and signature

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Informed Consent

I, the undersigned,…… declare that I have read the Patient Information Sheet, that I have understood its content, that I have been fully informed during the interview and that I have been given ample opportunity to ask questions about it.
Since I do not need to receive any further information, I request to take the test.

Date .....

Patient’s signature

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Physician's stamp and signature

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